



MFN2 gene

mitofusin 2

Normal Function

The *MFN2* gene provides instructions for making a protein called mitofusin 2. This protein helps determine the shape and structure (morphology) of mitochondria, the energy-producing centers within cells. Mitofusin 2 is made in many types of cells and tissues, including muscles, the spinal cord, and nerves that connect the brain and spinal cord to muscles (peripheral nerves). Within cells, mitofusin 2 is found in the outer membrane that surrounds mitochondria.

Mitochondria are dynamic structures that undergo changes in morphology through processes called fission (splitting into smaller pieces) and fusion (combining pieces). These changes in morphology are necessary for mitochondria to function properly. Mitofusin 2 helps to regulate the morphology of mitochondria by controlling the fusion process.

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

Researchers have identified approximately 50 *MFN2* gene mutations that cause a form of Charcot-Marie-Tooth disease known as type 2A. Almost all of these mutations change single protein building blocks (amino acids) in mitofusin 2. These genetic changes alter a critical region in mitofusin 2, and the protein cannot function properly. A few mutations create a premature stop signal in the instructions for making mitofusin 2. As a result, no protein is produced, or an abnormally small protein is made.

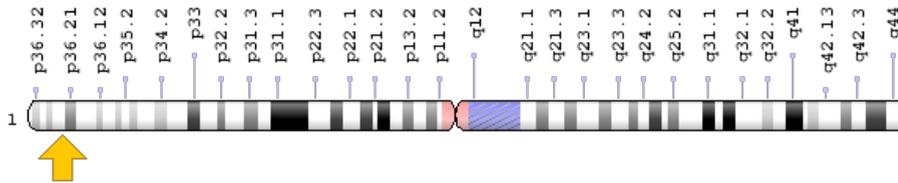
Several *MFN2* gene mutations cause a variant of type 2A Charcot-Marie-Tooth disease that affects vision. (This variant is also called hereditary motor and sensory neuropathy VI.) Vision loss is caused by the degeneration of the nerves that carry information from the eyes to the brain (optic atrophy). People with this variant usually experience severe symptoms of Charcot-Marie-Tooth disease that begin before age 10.

It is unclear how *MFN2* gene mutations lead to the nerve problems characteristic of type 2A Charcot-Marie-Tooth disease. Researchers suggest that mitochondria cannot fuse properly or move normally within the cell without functional mitofusin 2, which may disrupt the cell's energy supply. Nerve cells may be particularly sensitive to an interrupted supply of energy.

Chromosomal Location

Cytogenetic Location: 1p36.22, which is the short (p) arm of chromosome 1 at position 36.22

Molecular Location: base pairs 11,980,181 to 12,013,515 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CMT2A2
- CPRP1
- KIAA0214
- MARF
- MFN2_HUMAN
- mitochondrial assembly regulatory factor

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Dynamic mitochondrial reticulum
<https://www.ncbi.nlm.nih.gov/books/NBK26924/figure/A2605/>

GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 2A
<https://www.ncbi.nlm.nih.gov/books/NBK1511>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MFN2%5BTIAB%5D%29+OR+%28mitofusin+2%5BTIAB%5D%29%29+OR+%28%28MARF%5BTIAB%5D%29+OR+%28CPRP1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D>

OMIM

- MITOFUSIN 2
<http://omim.org/entry/608507>
- NEUROPATHY, HEREDITARY MOTOR AND SENSORY, TYPE VIA
<http://omim.org/entry/601152>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_MFN2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=MFN2%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16877
- Inherited Peripheral Neuropathies Mutation Database
<http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=32>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/9927>
- UniProt
<http://www.uniprot.org/uniprot/O95140>

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